

**WHAT IS CLAIMED:**

1. A method for screening subjects for genetic markers associated with autism, comprising:
  - 5 isolating a biological sample from a mammal; and testing the sample or genetic material isolated from the sample, for either a gene having a polymorphism or product thereof, which is a genetic marker for autism.
- 10 2. The method according to claim 1, wherein the biological sample is selected from the group consisting of blood, saliva, amniotic fluid, and tissue.
- 15 3. The method according to claim 2, wherein the biological sample is blood.
- 15 4. The method according to claim 1, wherein the mammal is a human.
- 20 5. The method according to claim 4, wherein the biological sample is isolated from developmentally disabled children.
- 20 6. The method according to claim 4, wherein the biological sample is isolated from parents or relatives of developmentally disabled children.
- 25 7. The method according to claim 4, wherein the biological sample is isolated from children and said method further comprises: early behavior training for children having genetic markers associated with autism.
- 30 8. The method according to claim 1, wherein the gene is selected from the group consisting of *HoxA1*, *HoxB1*, and *HoxD1*.
- 35 9. The method according to claim 8, wherein the polymorphism is located in the homeobox.
- 35 10. The method according to claim 8, wherein the gene is *HoxA1*.

11. The method according to claim 10, wherein the gene has a single base substitution resulting in an amino acid substitution.
- 5 12. The method according to claim 11, wherein the amino acid substitution is an arginine for a histidine.
13. The method according to claim 8, wherein the gene is *HoxB1*.
- 10 14. The method according to claim 12, wherein the gene has an insertion.
- 15 15. The method according to claim 14, wherein the insertion is 5'ACAGCGCCC-3'.
16. The method according to claim 8, wherein the mutated gene is *HoxD1*.
17. The method according to claim 1, wherein the gene has a polymorphism selected from the group consisting of a single base substitution resulting in an amino acid substitution, a single base substitution resulting in a translational stop, an insertion, a deletion, and a rearrangement.
- 20 18. The method according to claim 1, wherein the gene has a mutation in an exon.
- 25 19. The method according to claim 18, wherein the polymorphism alters the sequence of the polypeptide encoded by the gene.
20. The method according to claim 1, wherein the gene has a mutation in an intron.
- 30 21. The method according to claim 1, wherein the gene has a mutation in a promotor or regulatory region.
22. The method according to claim 1, wherein said testing is carried out by screening for a gene having a polymorphism.

23. The method according to claim 22, wherein said screening for mutated nucleic acids is carried out by a method selected from the group consisting of direct sequencing of nucleic acids, single strand polymorphism assay, restriction fragment length polymorphism assay, ligase chain reaction, enzymatic cleavage and southern hybridization.

24. The method according to claim 23, wherein said screening is carried out by direct sequencing of nucleic acids.

10 25. The method according to claim 23, wherein said screening is carried out by single strand polymorphism assay.

15 26. The method according to claim 23, wherein said screening is carried out by restriction fragment length polymorphism assay.

27. The method according to claim 23, wherein said screening is carried out by ligase chain reaction.

20 28. The method according to claim 23, wherein said screening is carried out by enzymatic cleavage.

29. The method according to claim 23, wherein said screening is carried out by southern hybridization.

25 30. The method according to claim 23, wherein the nucleic acid is a deoxyribonucleic acid.

30 31. The method according to claim 23, wherein the nucleic acid is a messenger ribonucleic acid.

32. The method according to claim 1, wherein said testing is carried out by screening for polypeptides resulting from said gene having a polymorphism.

35 33. The method according to claim 32, wherein said screening for the polypeptide resulting from said gene having a polymorphism is carried out by a

*j. 1/12* method selected from the group consisting of probing with antibodies specific to said polypeptide, measurement of the concentration of said polypeptide, and measuring the size of said polypeptide.

5        34.      The method according to claim 33, wherein said screening is carried out by probing with antibodies specific to said polypeptide.

10       35.      The method according to claim 33, wherein said screening is carried out by measuring the size of the polypeptides.

15       36.      An isolated nucleic acid molecule comprising a single base substitution at nucleotide 218 in SEQ ID No. 1,

16       or a fragment having at least 15 nucleotides encompassing said single base substitution.

20       37.      An isolated polypeptide encoded by the nucleic acid of claim 36.

25       38.      An antibody which binds to the isolated polypeptide according to claim 37 and which does not bind to the wild-type *HoxA1* protein of SEQ. ID. No. 2.

30       39.      An isolated nucleic acid molecule comprising an insertion between positions nucleotides 88 and 89 in SEQ. ID. No. 5,

31       or a fragment having at least 15 nucleotides encompassing said insertion.

35       40.      The isolated nucleic acid molecule according to claim 39, wherein the insertion is 5'-ACAGCCCCC-3'.

40       41.      An isolated polypeptide encoded by the nucleic acid of claim 39.

45       42.      An antibody which binds to the isolated polypeptide according to claim 41 and which does not bind to the wild-type *HoxB1* protein of SEQ. ID. No. 6.